Name:Ezeoke Chioma

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Assignment Title: IMMUNODEFICIENCY DISODERS

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Question

Immunodeficiency disorder is the absence or failure of normal function of one or more elements of the immune system. There are two major types of immunodeficiency disorders: PRIMARY AND SECONDARY.

1. Identify and briefly explain 5 primary immunodeficiency disorders

2. Identify and briefly explain 2 secondary immunodeficiency disoders

Answers

1. Primary immunodeficiency disorders include:

\* Wiscott-Aldrich syndrome

\* Severe combined immunodeficiency disease (SCID)

\* DiGeorge syndrome

\* Ataxia-Telangiectasis

\* Common variable immune deficiency (CVID)

\* Wiskott-Aldrich syndrome:

This is an X-linked disorder characterized by an altered cell surface glycoproteinstructure(CD43orsialophorin)commontolymphocytesand platelets. Its classic clinical features include a microcytic thrombocytope- nia that distinguishes this disorder from idiopathic thrombocytopenic purpura and other forms of normocytic thrombocytopenia. The immu- nologic findings are variable but usually include impaired humoral re- sponses to polysaccharide antigens and elevated serum IgA and IgE lev- els. Atopic dermatitis and recurrent pyogenic infections of the upper respiratory tract are common but variable associated clinical

Since the gene for sialophorin is located on chromosome 16, it was ap- parent that a genetic deficiency of sialophorin could not account for all the features of this X-linked disorder. More recently, the gene for Wiskott Aldrich syndrome has been identified and codes a large molecular weight signal transduction protein expressed on lymphocytes, megakaryocytes, spleen, and thymus. Techniques for both gene carrier detection and in utero diagnosis have been described.

\* Ataxia Telangiectasis:

Ataxia telangiectasia is an autosomal recessive disorder clinically characterized by progressive oculocutaneous telangiectasia, which becomes present between 3 and 6 years of age. Progressive cerebellar ataxia secondary to Purkinje’s cell degeneration typically results in loss of am- bulation by 10 to 12 years of age. Progeric changes of the skin and hair often are present. Immunologic features often include selective IgA and IgG2 subclass deficiency and depressed but not absent in vitro lympho- cyte responses. These patients also display exquisite toxicity to chemo- therapy and irradiation and should not receive these forms of diagnostic and therapeutic studies. This often poses difficultclinicaldecisionsas 15% of affected patients are estimated to develop malignancy. The ataxia tel- angiectasia gene was identified in 199528and has been referred to as a potential Rosetta stone of the human genome because of its wide-ranging roles. The postulated roles of this gene include detecting DNA damage, controlling immune system responses, preventing genomic rearrange- ments in malignancy, and preventing programmed cell death. It is be- lieved that u p to 1.4% of the population has one defective AT gene and the gene could account for up to 8% of all breast cancers. Treatment is limited to supportive care and no cure is available.Because the incidence of infection is so variable, bone marrow transplantation usually is not advised.

\* Thymic Hypoplasia(DiGeorge’s Syndrome):

Embryologists define this disorder as a developmental field defect involving the third and fourth branchial arches. These structures are needed for the normal development of the thymus, parathyroid glands, and the great vessels of the heart. Defective function of these organs ac- counts for the respective features of variable immunodeficiency,neonatal hypocalcemia secondary to hypoparathyroidism, and congenital cardiac defects. Abnormal ear, maxilla, and mandible development accounts for the characteristic facies of this disorder. Parathyroid and thymic deficiency also have been described in a disorder of peroxisomes known as Zellweger syndrome. When the DiGeorge’s syndrome phenotype is paired with cleft palate and learning disorders, the term velocardiofacialsyndrome is used. Diagnosis in newborns should be suspected when hypocalcemia is paired with such relatively uncommon cardiac anomalies as an interrupted aortic arch type B, right-sided aortic arch or truncus arteriosus. Ninety percent of DiGeorge’s syndrome patients have associated chromosomedisorders, with the majority involving chromosome 22. The immunologic findings are variable, and spontaneous improvement in the immunologic defect has been described. In its severest form,bone marrow transplantation may be required.

•Severe Combined Immunodeficiency (SCID):

This term is used to describe a group of disorders that involve both the B and T cells. Most of the B-cell abnormalities appear secondary to the lack of T-cell help, which points again to the critical role of the T cell as the leader of the immune system. The consequence of absent T-cell function is opportunistic infections and overwhelming septicemia, with such pathogens as Pneumocystis carinii, vaccinia, varicella and measles. These infants also are at risk for lethal GVHD if given transfusions with nonirradiated blood products. Lymphopenia is a common finding in X- linked SCID, but may be masked by the presence of maternal T cells, which have crossed the placenta and engraftment in the infant. Rarer forms of SCID may present with lymphocytosissecondary to proliferation of clones of dysfunctional T cells. The severity of these immunologic dis- orders has made them logical, early candidates for such advances in treat- ment as bone marrow transplantation and gene replacement therapy. Early diagnosis and availability of matched donors for bone marrow transplantation remains the most important prognostic factor for this group of severe disorders. SCID has become recognized as a heteroge- neous group of disorders of stem cell and thymic differentiation,surface receptor expression, cellular signaling and enzyme deficiency.

•Common Variable Immune Deficiency(CVID):

CVID is a diagnosis of exclusion, as the genetic basis remains un- known. It is clinically indistinguishable from the other primary B-cell dis- orders and shares features of hypogammaglobulinemia, recurrent pyo- genic infection,and impaired antibody responses. In contrast to XLA, the onset usually is in the second or third decade of life. The clinical, immu- nologic, and genetic diversity of CVID suggests that this may represent a common clinical framework for several genetically distinct disorders? In addition to the features of impaired humoral immunity, up to 50% of CVID patients exhibit some alteration of T-cell function. This has included low CD4/CD8 ratios, low expression of T-cell activation molecules, ab- normal responses to T-cell mitogens, and reduced lymphokine produc- tion. Passive replacement therapy with high dose IVIG has been shown to reduce the incidence of pyogenic infections and is a standard of care for this group of illness.

2. Secondary immunodeficiency (SID)

SIDs are more common than PIDs and are the result of a primary illness, such as HIV, or other external factor such as malnutrition or some drug regimens. Most SIDs can be resolved by treating the primary condition.

Examples of secondary immunodeficiency disorders

\* Malnutrition – Protein-calorie malnutrition is the biggest global cause of SIDs which can affect up to 50% of the population in some communities in the developing world.vii T cell numbers and function decrease in proportion to levels of protein deficiency, which leaves the patient particularly susceptible to diarrhoea and respiratory tract infections. This form of immunodeficiency will usually resolve if the malnutrition is treated.

\* Chronic infections – There are a number of chronic infections which can lead to SID disorders, the most common of which is acquired immune deficiency syndrome (AIDS), resulting from HIV infection. The virus attacks CD4+ T cells, a type of white blood cell that plays a critical role in preventing infection, and gradually depletes their numbers. Once the T cell count is less than 200 cells per ml of blood, symptoms of AIDS begin to manifest and the patient is at high risk of recurrent infections that will eventually lead to death. Anti-viral therapies, such as the HAART regimen (Highly Active Antiretroviral Therapy), allow the T cell population a chance to recover and resume normal function. These drugs have had a huge impact on increasing the life expectancy for HIV/AIDS patients and improving their quality of life. Prior to the introduction of HAART, patients with HIV diagnosed at age 20 had an average of 10 years before developing AIDS. Nowadays on average, patients diagnosed at age 20 can expect to live well into their 60s.viii However, these drugs must be taken every day for life as they are not curative, and are only available to patients and healthcare systems that can afford them.